

What is claimed is:

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1. A method for diagnosing or aiding in the diagnosis of a vascular disease or disorder in a subject comprising the steps of determining the PLOD2 genetic profile of the  
5 subject, thereby diagnosing or aiding in the diagnosis of a vascular disease or disorder.
2. The method of claim 1, wherein determining the subject's PLOD2 genetic profile comprises determining the identity of the nucleotide present at nucleotide position 147472 of SEQ ID NO:1, or the complement thereof.  
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3. The method of claim 1, wherein the vascular disease is myocardial infarction.
4. The method of claim 1, wherein the vascular disease is coronary artery disease.  
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5. A method for predicting the likelihood that a subject will or will not develop a vascular disease or disorder comprising the steps of determining the PLOD2 genetic profile of the subject, thereby predicting the likelihood that a subject will or will not develop a vascular disease or disorder.  
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6. The method of claim 5, wherein determining the subject's PLOD2 genetic profile comprises determining the identity of the nucleotide present at nucleotide position 147472 of SEQ ID NO:1, or the complement thereof.  
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7. The method of claim 5, wherein the vascular disease is myocardial infarction.
8. The method of claim 5, wherein the vascular disease is coronary artery disease.

9. A method of diagnosing or aiding in the diagnosis of a vascular disease in a subject comprising the steps of determining the nucleotide present at nucleotide position 147472 of the PLOD2 gene, wherein the presence of two copies of an adenine allele at  
5 nucleotide position 147472 of the PLOD2 gene, or the complement thereof, is indicative of decreased likelihood of a vascular disease in the subject as compared with a subject having any other combination of alleles at this locus.

10. The method of claim 9, wherein determining said nucleotides comprises  
10 obtaining a nucleic acid sample from the subject.

11. The method of claim 9, wherein the PLOD2 gene has the nucleotide sequence of SEQ ID NO:1, or a portion thereof.

15 12. The method of claim 9, wherein the vascular disease is selected from the group consisting of atherosclerosis, coronary artery disease, myocardial infarction, ischemia, stroke, peripheral vascular diseases, venous thromboembolism and pulmonary embolism.

20 13. The method of claim 12, wherein the vascular disease is myocardial infarction.

14. The method of claim 12, wherein the vascular disease is coronary artery disease.

25 15. A method for predicting the likelihood that a subject will or will not develop a vascular disease, comprising the steps of determining the nucleotide present at nucleotide position 147472 of the PLOD2 gene, wherein the presence of two copies of an adenine allele at nucleotide position 147472 of the PLOD2 gene, or the complement thereof, is indicative of decreased likelihood of the subject developing a vascular disease as compared with a

subject having any other of alleles at this locus.

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16. The method of claim 15, wherein determining said nucleotides comprises obtaining a nucleic acid sample from the subject.

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17. The method of claim 15, wherein the PLOD2 gene has the nucleotide sequence of SEQ ID NO:1, or a portion thereof.

18. The method of claim 15, wherein the vascular disease is selected from the group consisting of atherosclerosis, coronary artery disease, myocardial infarction, ischemia, stroke, peripheral vascular diseases, venous thromboembolism and pulmonary embolism.

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19. The method of claim 18, wherein the vascular disease is myocardial infarction.

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20. The method of claim 19, wherein the vascular disease is coronary artery disease.

21. A computer readable medium for storing instructions for performing a computer implemented method for determining whether or not a subject has a predisposition to a vascular disease or disorder, said instructions comprising the functionality of:

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obtaining information from the subject indicative of the presence or absence of the polymorphic region of a PLOD2 gene, and

based on the presence or absence of the polymorphic region of a PLOD2 gene, determining whether or not the subject has a predisposition to a vascular disease or disorder.

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22. A computer readable medium for storing instructions for performing a computer implemented method for identifying a predisposition to a vascular disease or disorder, said instructions comprising the functionality of:

obtaining information regarding the presence or absence of the polymorphic region of a PLOD2 gene, and

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based on the presence or absence of the polymorphic region of a PLOD2 gene, identifying a predisposition to a vascular disease or disorder.

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23. An electronic system comprising a processor for determining whether or not a subject has a predisposition to a vascular disease or disorder, said processor implementing the functionality of:

obtaining information from the subject indicative of the presence or absence of the polymorphic region of a PLOD2 gene, and

based on the presence or absence of the polymorphic region of a PLOD2 gene, determining whether or not the subject has the predisposition to a vascular disease or disorder.

15 24. An electronic system comprising a processor for performing a method for identifying a predisposition to a vascular disease or disorder in a subject, said processor implementing the functionality of:

obtaining information from the subject indicative of the presence or absence of the polymorphic region of a PLOD2 gene, and

20 based on the presence or absence of the polymorphic region of a PLOD2 gene, performing a method for identifying a predisposition to a vascular disease or disorder associated with the polymorphic region.

25 25. The electronic system of claims 23 or 24, wherein said processor further implements the functionality of receiving phenotypic information associated with the subject.

26. The electronic system of claims 23 or 24, wherein said processor further implements the functionality of acquiring from a network phenotypic information associated with the subject.

27. A network system for identifying a predisposition to a vascular disease or disorder in response to information submitted by an individual, said system comprising means for:

5 receiving data from the individual regarding the presence or absence of the polymorphic region of a PLOD2 gene, and

based on the presence or absence of the polymorphic region, determining whether or not the subject has the predisposition to the vascular disease or disorder associated with the polymorphic region.

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28. A network system for identifying whether or not a subject has a predisposition to a vascular disease or disorder, said system comprising means for:

receiving information from the subject regarding the polymorphic region of a PLOD2 gene,

15 receiving phenotypic information associated with the subject,  
acquiring additional information from the network, and

based on one or more of the phenotypic information, the polymorphic region, and the acquired information, determining whether or not the subject has a pre-disposition to a vascular disease or disorder associated with a polymorphic region of a PLOD2 gene.

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29. The system of claims 27 and 28, wherein the network system comprises a server and a work station operatively connected to said server via the network.

30. A method for determining the identity an allelic variant of a polymorphic  
25 region of a PLOD2 gene in a nucleic acid obtained from a subject, comprising contacting a sample nucleic acid from the subject with a probe or primer having a sequence which is complementary to a PLOD2 gene sequence, wherein the sample comprises a PLOD2 gene sequence, thereby determining the identity of an of the allelic variant.

31. The method of claim 30, wherein the probes or primers are capable of hybridizing to an allelic variant of a polymorphic region of the PLOD2 gene, and wherein the allelic variant differs from the reference sequence set forth in of SEQ ID NO:1.

5           32. The method of claim 31, wherein determining the identity of the allelic variant comprises determining the identity of at least one nucleotide of the polymorphic region of a PLOD2 gene.

10           33. The method of claim 32, wherein determining the identity of the allelic variant consists of determining the nucleotide content of the polymorphic region.

34. The method of claim 32, wherein determining the nucleotide content comprises sequencing the nucleotide sequence.

15           35. The method of claim 32, wherein determining the identity of the allelic variant comprises performing a restriction enzyme site analysis.

36. The method of claim 32, wherein determining the identity of the allelic variant is carried out by single-stranded conformation polymorphism.

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37. The method of claim 32, wherein determining the identity of the allelic variant is carried out by allele specific hybridization.

38. The method of claim 32, wherein determining the identity of the allelic variant is carried out by primer specific extension.

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39. The method of claim 32, wherein determining the identity of the allelic variant is carried out by an oligonucleotide ligation assay.

40. The method of claim 32, wherein the probe or primer comprises a nucleotide sequence from about 15 to about 30 nucleotides.

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5 41. An Internet-based method for assessing a subject's risk for vascular disease, the method comprising:

- a) analyzing biological information from a subject indicative of the presence or absence of a polymorphic region of PLOD2;
- b) providing results of the analysis to the subject via the Internet, wherein the presence of a polymorphic region of PLOD2 indicates a decreased risk for vascular
- 10 disease.

42. A method of assessing a subject's risk for vascular disease, the method comprising:

- a) obtaining biological information from the individual;
- 15 b) analyzing the information to obtain the subject's PLOD2 genetic profile;
- c) representing the PLOD2 genetic profile information as digital genetic profile data;
- d) electronically processing the PLOD2 digital genetic profile data to
- 20 generate a risk assessment report for vascular disease, wherein the presence of a polymorphic region of PLOD2 indicates a decreased risk for vascular disease; and
- e) displaying the risk assessment report on an output device.

25 43. A method of assessing a subject's risk for vascular disease, the method comprising:

- a) obtaining the subject's PLOD2 genetic profile information as digital genetic profile data;

- b) electronically processing the PLOD2 digital genetic profile data to generate a risk assessment report for vascular disease, wherein the presence of a polymorphic region of PLOD2 indicates a decreased risk for vascular disease; and
- c) displaying the risk assessment report on an output device.

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44. The method of claims 42 or 43, further comprising the step of using the risk assessment report to provide medical advice.

45. The method of claims 42 or 43, wherein additional health information is  
10 provided.

46. The method of claim 45, wherein the additional health information comprises information regarding one or more of age, sex, ethnic origin, diet, sibling health, parental health, clinical symptoms, personal health history, blood test data, weight, and alcohol use,  
15 drug use, nicotine use, and blood pressure.

47. The method of claim 43, wherein the PLOD2 digital genetic profile data are transmitted via a communications network to a medical information system for processing.

20 48. The method of claim 47, wherein the communications network is the Internet.

49. A medical information system for assessing a subject's risk for vascular disease comprising:

- a) means for obtaining biological information from the individual to  
25 obtain a PLOD2 genetic profile;
- b) means for representing the PLOD2 genetic profile as digital molecular data;
- c) means for electronically processing the PLOD2 digital genetic profile to generate a risk assessment report for vascular disease; and



d) means for displaying the risk assessment report on an output device, wherein the presence of a polymorphic region of PLOD2 indicates a decreased risk for vascular disease.

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5           50. A medical information system for assessing a subject's risk for vascular disease comprising:

a) means for representing the subject's PLOD2 genetic profile data as digital molecular data;

b) means for electronically processing the PLOD2 digital genetic profile  
10 to generate a risk assessment report for vascular disease; and

c) means for displaying the risk assessment report on an output device, wherein the presence of a polymorphic region of PLOD2 indicates a decreased risk for vascular disease.

15           51. A computerized method of providing medical advice to a subject comprising:

a) analyzing biological information from a subject to determine the subject's PLOD2 genetic profile;

b) based on the subject's PLOD2 genetic profile, determining the subject's risk for vascular disease;

20 c) based on the subject's risk for vascular disease, electronically providing medical advice to the subject.

52. A computerized method of providing medical advice to a subject comprising:

a) based on the subject's PLOD2 genetic profile, determining the  
25 subject's risk for vascular disease;

b) based on the subject's risk for vascular disease, electronically providing medical advice to the subject.

53. The method of any of claims 51 or 52, wherein the medical advice comprises

one or more of the group consisting of further diagnostic evaluation, administration of medication, or lifestyle change.

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54. The method of claims 51 or 52, wherein additional health information is  
5 obtained from the subject.

55. The method of claim 54, wherein the additional health information comprises  
information regarding one or more of age, sex, ethnic origin, diet, sibling health, parental  
health, clinical symptoms, personal health history, blood test data, weight, and alcohol use,  
10 drug use, nicotine use, and blood pressure.

56. A method for self-assessing risk for a vascular disease comprising  
a) providing biological information for genetic analysis;  
b) accessing an electronic output device displaying results of the genetic  
15 analysis, thereby self-assessing risk for a vascular disease, wherein the presence of a  
polymorphic region of PLOD2 indicates a decreased risk for vascular disease.

57. A method for self-assessing risk for a vascular disease comprising accessing  
an electronic output device displaying results of a genetic analysis of a biological sample,  
20 wherein the presence of a polymorphic region of PLOD2 indicates a decreased risk for  
vascular disease, thereby self-assessing risk for a vascular disease.

58. A method of self-assessing risk for vascular disease, the method comprising  
a) providing biological information;  
25 b) accessing PLOD2 digital genetic profile data obtained from the  
biological information, the PLOD2 digital genetic profile data being displayed via an  
output device, wherein the presence of a polymorphic region of PLOD2 indicates a  
decreased risk for vascular disease.

59. A method of self-assessing risk for vascular disease, the method comprising accessing PLOD2 digital genetic profile data obtained from biological information, the PLOD2 digital genetic profile data being displayed via an output device, wherein the presence of a polymorphic region of PLOD2 indicates a decreased risk for vascular disease.

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60. The method of claims 58 or 59, wherein the electronic output device is accessed via the Internet.

61. The method of claims 58 or 59, wherein additional health information is provided.

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62. The method of claim 61, wherein the additional health information comprises information regarding one or more of age, sex, ethnic origin, diet, sibling health, parental health, clinical symptoms, personal health history, blood test data, weight, and alcohol use, drug use, nicotine use, and blood pressure.

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63. The method of any of claims 56, 57, 58, or 59, wherein the biological information is obtained from a sample from an individual at a laboratory company.

64. The method of claim 63, wherein the laboratory company processes the biological sample to obtain PLOD2 genetic profile data, represents at least some of the PLOD2 genetic profile data as digital genetic profile data, and transmits the PLOD2 digital genetic profile data via a communications network to a medical information system for processing.

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65. The method of any of claims 56, 57, 58, or 59, wherein the biological information is obtained from a sample from an individual at a draw station, wherein the draw station processes the biological sample to obtain PLOD2 genetic profile data, and transfers the data to a laboratory company.

66. The method of claim 65, wherein the laboratory company represents at least  
some of the PLOD2 genetic profile data as digital genetic profile data, and transmits the  
PLOD2 digital genetic profile data via a communications network to a medical information  
5 system for processing.

67. A method for a health care provider to generate a personal health assessment  
report for an individual, the method comprising counseling the individual to provide a  
biological sample; authorizing a draw station to take a biological sample from the individual  
10 and transmit molecular information from the sample to a laboratory company, wherein the  
molecular information comprises the presence or absence of a polymorphic region of  
PLOD2; requesting the laboratory company to provide digital molecular data corresponding  
to the molecular information to a medical information system to electronically process the  
digital molecular data and digital health data obtained from the individual to generate a  
15 health assessment report; receiving the health assessment report from the medical  
information system; and providing the health assessment report to the individual.

68. A method for a health care provider to generate a personal health assessment  
report for an individual, the method comprising requesting a laboratory company to provide  
20 digital molecular data corresponding to the molecular information derived from a biological  
sample from the individual to a medical information system to electronically process the  
digital molecular data and digital health data obtained to generate a health assessment report;  
receiving the health assessment report from the medical information system; and providing  
the health assessment report to the individual.

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69. A method of assessing the health of an individual, the method comprising:  
obtaining health information from the individual using an input device; representing at least  
some of the health information as digital health data; obtaining biological information from  
the individual, wherein the information comprises the presence or absence of a polymorphic

region of PLOD2; representing at least some of the information as digital molecular data; electronically processing the digital molecular data and digital health data to generate a health assessment report; and displaying the health assessment report on an output device.

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5           70.    The method of claim 69, wherein electronically processing the digital molecular data and digital health data to generate a health assessment report comprises using the digital molecular data and digital health data as inputs for an algorithm or a rule-based system that determines whether the individual is at risk for a specific disorder.

10           71.    The method of claim 69, wherein the individual has or is at risk of developing vascular disease, and wherein electronically processing the digital molecular data and digital health data to generate a health assessment report comprises using the digital molecular data and digital health data as inputs for an algorithm or a rule-based system that determines the individual's prognosis.

15           72.    The method of claim 69, wherein electronically processing the digital molecular data and digital health data comprises using the digital molecular data and digital health data as inputs for an algorithm or a rule-based system based on one or more databases comprising stored digital molecular data and/or digital health data relating to one or more  
20 disorders.

          73.    The method of claim 69, wherein electronically processing the digital molecular data and digital health data comprises using the digital molecular data and digital health data as inputs for an algorithm or a rule-based system based on one or more databases  
25 comprising (i) stored digital molecular data and/or digital health data from a plurality of healthy individuals, and (ii) stored digital molecular data and/or digital health data from one or more pluralities of unhealthy individuals, each plurality of individuals having a specific disorder.

74. The method of either of claims 72 or 73, wherein at least one of the databases is a public database.

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5 75. The method of claim 69, wherein the digital health data and digital molecular data are transmitted via a communications network to a medical information system for processing.

76. The method of claim 75, wherein the communications network is the Internet.

10 77. The method of claim 75, wherein the input device is a keyboard, touch screen, hand-held device, telephone, wireless input device, or interactive page on a website.

78. The method of claim 75, wherein the health assessment report comprises a digital molecular profile of the individual.

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79. The method of claim 75, wherein the health assessment report comprises a digital health profile of the individual.

80. The method of claim 75, wherein the molecular data comprises nucleic acid sequence data, and the molecular profile comprises a genetic profile.

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81. The method of claim 75, wherein the molecular data comprises protein sequence data, and the molecular profile comprises a proteomic profile.

25 82. The method of claim 75, wherein the molecular data comprises information regarding one or more of the absence, presence, or level, of one or more specific proteins, polypeptides, chemicals, cells, organisms, or compounds in the individual's biological sample.

83. The method of claim 75, wherein the health information comprises information relating to one or more of age, sex, ethnic origin, diet, sibling health, parental health, clinical symptoms, personal health history, blood test data, weight, and alcohol use, drug use, nicotine use, and blood pressure.

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84. The method of claim 75, wherein the health information comprises current and historical health information.

85. The method of claim 75, further comprising obtaining a second set of  
10 biological information at a time after obtaining the first set of biological information;  
processing the second set of biological information to obtain a second set of information;  
representing at least some of the second set of information as digital second molecular data;  
and processing the molecular data and second molecular data to generate a health assessment  
report.

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86. The method of claim 85, further comprising obtaining second health  
information at a time after obtaining the health information; representing at least some of the  
second health information as digital second health data and processing the molecular data,  
health data, second molecular data, and second health data to generate a health assessment  
20 report.

87. The method of claim 75, wherein the health assessment report provides  
information about the individual's predisposition for vascular disease and options for risk  
reduction.

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88. The method of claim 87, wherein the options for risk reduction comprise one  
or more of diet, exercise, one or more vitamins, one or more drugs, cessation of nicotine use,  
and cessation of alcohol use.

89. The method of claim 75, wherein the health assessment report provides information about treatment options for a particular disorder.

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90. The method of claim 89, wherein the treatment options comprise one or more of diet, one or more drugs, physical therapy, and surgery.

91. The method of claim 75, wherein the health assessment report provides information about the efficacy of a particular treatment regimen and options for therapy adjustment.

92. The method of claim 75, further comprising storing the molecular data.

93. The method of claim 92, further comprising building a database of stored molecular data from a plurality of individuals.

94. The method of claim 75, further comprising storing the molecular data and health data.

95. The method of claim 94, further comprising building a database of stored molecular data and health data from a plurality of individuals.

96. The method of claim 95, further comprising building a database of stored digital molecular data and/or digital health data from a plurality of healthy individuals, and stored digital molecular data and/or digital health data from one or more pluralities of unhealthy individuals, each plurality of individuals having a specific disorder.